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Overview

The more widespread availability of early intervention and growing evidence of its efficacy for children with autism spectrum disorders (ASDs) have led to a recent emphasis on screening and early diagnosis. ASDs, which include autism, Asperger's disorder, and pervasive developmental disorder-not otherwise specified (PDD-NOS), currently have no biological marker. They are defined by behavior, including the absence of normal social engagement and communication, and the presence of unusual repetitive behaviors. Screening and diagnosis require accurate behavioral reports or observations. In this article, we will discuss screening and diagnosis, with reference to current research on identification of children with ASD at age 2 years or younger, through population (level 1) and focused (level 2) screening, and then with standard diagnostic methods.

Screening for ASD: Current Findings

The Checklist for Autism in Toddlers

Figure 1 lists the various screening tools used for identifying ASD. Population screening for ASD was initially attempted in the United Kingdom using the Checklist for Autism in Toddlers (CHAT),[1] which emphasized joint attention and imagination. Nurses visited 18-month-old children at home and administered the CHAT activities to them and a series of questions to their parents. Most children classified by the CHAT were later diagnosed with autism. However, upon follow-up, it became evident that the CHAT had missed more than three quarters of children who eventually received ASD diagnoses. Furthermore, children with suspected developmental disabilities had been eliminated even before the screening, so it was unclear how the overlap between autism and mental retardation would be treated.

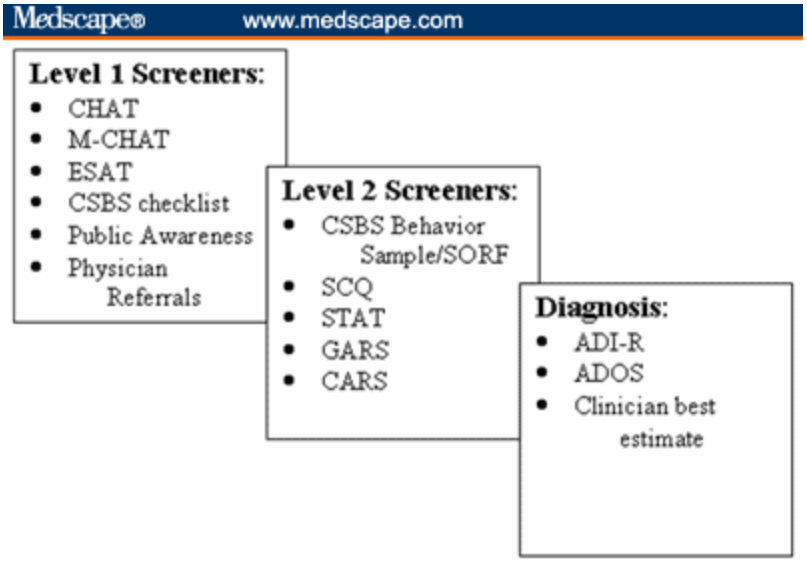

Figure 1.

Levels of screening and diagnosis for children with ASD at age 2 years or younger.

Modified CHAT

In 2001, Robins and colleagues[2] published results of a study of a modified version of the CHAT, the M-CHAT, which they administered to children 24 months of age who were recruited from pediatric practices and special education programs in the United States. This modified version eliminated the nurse-administered component and added

more parent questions. Like the CHAT, the M-CHAT identified children with autism at age 2 years, but because most of these children had been recruited from special education programs, a larger population-based study of the M-CHAT is necessary (and is underway).

The Early Screening for Autistic Traits

The Early Screening for Autistic Traits (ESAT),[3] puts a greater emphasis on play and less on joint attention than the previous instruments. Results of investigations of this tool indicated that children with high scores on the ESAT were likely to have developmental problems, but for children younger than 24 months, the measure did not differentiate well between ASD and non-ASD. Moreover, like the CHAT and M-CHAT, it also missed many children who later received ASD diagnoses.[3,4] Nevertheless, despite the ESAT's poor sensitivity, the study increased public awareness and provided a simple way to pursue referrals, and thus early identification increased.

The Communication Symbols Behavior Scales

The Communication Symbols Behavior Scales (CSBS)[5] is a 1-page parent questionnaire designed to identify communication disorders, rather than ASD in particular, in 6- to 24-month-old children. If children screen positive, a direct assessment (the Behavior Sample) and an additional parent-caregiver questionnaire are administered. The Scale of Red Flags (SORF)[6] for autism was developed for scoring the Behavior Sample as a level 2 screener. It successfully identified most children with language delay as having or not having autism.

The Social Communication Questionnaire

The Social Communication Questionnaire (SCQ)[7] is a parent-caregiver questionnaire intended to identify potential participants for autism studies. It was normed on older children and adults. Research by Corsello and colleagues (unpublished data, 2005) has indicated that if the cut-off is adjusted to require fewer endorsed items, the SCQ worked well for children as young as 3 years old.[8] However, the children studied had already been referred for services, so the appropriateness of the SCQ with the general population is unclear, as is its usefulness with children younger than 3 years old.

STAT, GARS, and CARS

The Screening Test for Autism in Two Year Olds (STAT)[9] is directly administered to the child by a trained examiner. It discriminates well between children with ASD and children with other developmental disorders. It is intended for children already suspected of having ASD.

Two other well-known scales are the Gilliam Autism Rating Scale (GARS)[10,11] and the Childhood Autism Rating Scale (CARS).[12] Both of these tools are primarily intended for level 2 screening but they are sometimes mistakenly used as diagnostic instruments. Both instruments are not commonly used with young children.

Screening for ASD: Other Identifying Characteristics

Overall, screening for autism has been successful in some respects and not in others. Poor scores on any of the instruments suggest that a child is at high risk for having an ASD. A primary care physician concerned about possible ASD might administer the CSBS screener or M-CHAT and then refer a high-scoring child for administration of a second level screener (such as the STAT or the CSBS Behavior Sample/Caregiver Questionnaire).

The difficulty lies in the children not identified by level 1 screeners, even though descriptive studies have found important behavioral differences in children approaching their first birthdays.[13,14] Retrospective data from studies of children with autism identified at age 2 or 3 years suggest several other factors that primary caregivers may want to

take into account when identifying children at risk for ASD.[15]

First, roughly 20% of children with ASD experience regressions, usually between 13 and 24 months, during which they stop talking and become more withdrawn.[15] Even more children may appear to "worsen," not obviously losing skills but showing subtle qualitative deteriorations in social responsiveness. These children may not "fail" the level 1 screening tests if their parents answer in terms of the skills they had at their best. However, any parental indication that a child has stopped talking, using gestures, or engaging in other social behaviors for longer than a few weeks should be taken very seriously.

Second, caregivers rarely express concern that their baby is in a "world of his own" unless something is wrong developmentally.[12] Thus, if a parent expresses worries about a child's social engagement, the physician should observe the child's nonverbal communication and social interactions and schedule another visit in a few weeks. If any doubts remain, the physician should refer the child to a clinic with staff experienced in identifying very young children at risk for ASDs.

Third, many children with ASD have significant delays in receptive and expressive language. A child who has met motor milestones and has intact hearing but who does not understand any words out of context warrants concern for possible ASD. Likewise, a child who has a vocabulary of letter names or colors but does not say "mama" or "dada" by age 2 also merits attention for possible ASD. In both of these cases, a referral to a speech-language pathologist and an interdisciplinary autism center could initiate treatment, even if ASD is later found not to be the most appropriate diagnosis.

Finally, researchers are now studying the early development of ASD prospectively, following infant siblings of children with autism because they are at risk (about 1/10 to 1/20) for developing ASD. Siblings who later develop autism appear to show early differences in social and communication behaviors.[11] The Autism Observation Scale for Infants (AOSI) [16] measures behaviors such as eye tracking and attention shifts to identify children who will later be diagnosed with autism. It is hoped that eventually these methods will provide clinical information as well.

Early Diagnosis

A number of prospective studies have suggested that, when using standard diagnostic measures such as the Autism Diagnostic Interview-Revised (ADI-R)[17] and the Autism Diagnostic Observation Scale (ADOS),[18] children can be accurately diagnosed with ASD when as young as 24 months old. These instruments produce reliable information about even very young children. However, their diagnostic thresholds cannot be applied to children with nonverbal mental ages below 15 months on the ADOS or 18 months on the ADI-R, as doing so would result in the misdiagnosis of a substantial minority of children who do not have autism but are at very low levels developmentally.

These findings lead to 4 summary points about early diagnosis. First, a competent measure of nonverbal problem-solving and language level (such as the Mullen Scales of Early Learning[19] or Bayley Scale of Infant Development, 3rd edition[20]) is crucial for interpreting information relevant to diagnosis of ASD in very young children.

Second, in several longitudinal studies, diagnoses made by experienced clinicians of children at age 2 years were the most powerful predictors of outcome.[9,12,21] In another study, more false positive errors (saying that a child did not have ASD when he did) occurred during the first months of a study than in the remaining 2 years (Lord, unpublished data, 2005). Though very experienced with older preschool children, the examiners lacked experience diagnosing very young children. Thus, experience with diagnosing very young children with ASD is critical.

Third, when diagnoses were confirmed across several sources (for example, the ADI-R, ADOS, and clinicians' best estimate), diagnoses made with 2 or more sources were much more predictive of later diagnosis than diagnoses based on only 1 or 2 sources of information (Lord, unpublished data, 2005) (Figure 2).

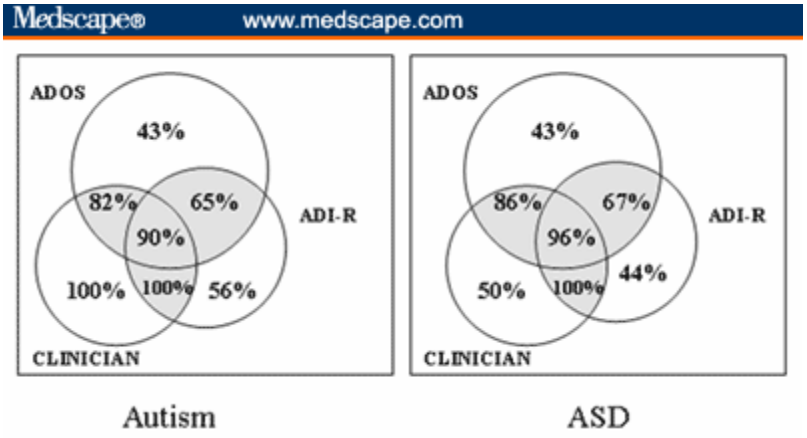

Figure 2.

Percent of age 2 diagnostic combinations and best-estimate judgments accurately predicting diagnosis at age 9.

Fourth, when age 2 language level was taken into account, repetitive behaviors were a stronger predictor of a stable diagnosis from ages 2 to 9 years than social behavior (Lord, unpublished data, 2005). Earlier studies indicated that not all children who meet autism criteria at 3 or older had repetitive behaviors at 2 years of age.[12,22,23] This suggests that repetitive behaviors at very young ages are not a necessary component of ASD, but if they are present, it is significant.

The ADI-R and ADOS are diagnostic instruments and are different from screeners. They are more time-consuming (and more expensive), require administration by a trained examiner, and provide information that can be used to identify educational objectives and make treatment plans.

Early Diagnosis of ASD: Summary and Conclusions

Altogether, an enormous amount of information about young children with autism and ASD has been acquired in the past decade. This information indicates that children can be diagnosed with autism or ASD at 2 years of age. The exception, however, is when the child has skills below the 15 month level, in which case the distinction between moderate or severe developmental delays with and without autism may be unreliable. Nonetheless, these children can benefit from assessment and ensuing interventions, even if their diagnoses change in the following years.

It is clear that screeners can identify children most likely to have autism or ASD from populations of children already referred for delays. Descriptive research has also shown that children with autism or ASD have fewer prosocial behaviors even as they reach their first birthdays,[10,11] but to date these findings have not been put into practice. Nevertheless, there are numerous “red flags” for autism that physicians can attend to, such as the occurrence of a regression, parent concerns about a child “being in a world of his own,” and severely delayed receptive language as long as hearing is intact. Accompanied by accessible information and places for referral, awareness itself may make the biggest difference for early identification.

References

1. Baron-Cohen S, Allen J, Gillberg C. Can autism be detected at 18 months? The needle, the haystack, and the CHAT. Br J Psychiatry. 1992;161:839-843. Abstract

2. Robins DL, Fein D, Barton ML, Green JA. The Modified Checklist for Autism in Toddlers: an initial study investigating the early detection of autism and pervasive developmental disorders. J Autism Dev Disord. 2001;31:131-144. Abstract

3. Willemsen-Swinkels SHN, Dietz C, Van Daalen E, et al. Screening for autistic spectrum disorders in children aged 14 to 15 months: the development of the Early Screening of Autistic Traits questionnaire. J Autism Dev Disord. In press.

4. Buitelaar JK, Willemsen-Swinkels S, Daalen E, et al. The screening instrument for the early detection of autism. Program and abstracts of the American Academy of Child and Adolescent Psychiatry 47th Annual Meeting; October 24-29, 2000; New York, NY.

5. Wetherby A. Communication and Symbolic Behavior Scales Developmental Profile, Preliminary Normed Edition. Baltimore, Md: Paul H. Brookes Publishing; 2001.

6. Wetherby AM, Woods J. Systematic observation of red flags for autism spectrum disorders in young children: unpublished manual. Tallahassee, Fl: Florida State University; 2002.

7. Rutter M, Bailey A, Lord C, Berument S K. Social Communication Questionnaire. Los Angeles, Calif: Western Psychological Services; 2003.

8. A, Ware J, Lord C. Social Communication Questionnaire (SCQ). Program and abstracts of the International Meeting for Autism Research (IMFAR); November 1-2, 2002; Orlando, Florida.

9. Stone WL, Coonrod EE, Ousley OY. Screening Tool for Autism Two-Year-Olds (STAT): development and preliminary data. J Autism Dev Disord. 2000;30:607-612. Abstract

10. Gilliam JE. Gilliam Autism Rating Scale. Austin, Tex: Pro-Ed; 1995.

11. South M, Williams B, McMahon W, et al. Utility of the Gilliam Autism Rating Scale in research and clinical populations. J Autism Dev Disord. 2002;32:593-599. Abstract

12. Schopler E, Reichler RJ, DeVellis RF, Daly K. Toward objective classification of childhood autism: Childhood Autism Rating Scale (CARS). J Autism Dev Disord. 1980;10:91-103. Abstract

13. Werner E, Dawson G, Osterling J, Dinno N. Brief report: recognition of autism spectrum disorder before one year of age: a retrospective study based on home videotapes. J Autism Dev Disord. 2000;30:157-162. Abstract

14. Zwaigenbaum L, Bryson S, Rogers T, et al. Behavioral manifestations of autism in the first year of life. Int J Dev Neurosci. 2005;23:143-152. Abstract

15. Hanson E, Sullivan N, Thurm Lord C. Follow-up of two-year-olds referred for possible autism. J Child Psychol Psychiatry Allied Disciplines. 1995;36:1365-1382.

16. Bryson SE, McDermott C, Rombough V, Zwaigenbaum L. The Autism Observational Scale for Infants: scale development and reliability data. J Autism Dev Disord. In press.

17. LeCouteur A, Lord C, Rutter M. The Autism Diagnostic Interview - Revised (ADI-R). Los Angeles, Calif: Western Psychological Services; 2003.

18. Lord C, Rutter M, DiLavore PC, Risi S. Autism Diagnostic Observation Schedule. Los Angeles, Calif: Western Psychological Services; 2002.

19. Mullen E. Mullen Scales of Early Learning. Circle Pines, Mn: American Guidance Service Inc; 1995.

20. Bayley N. Bayley Scales of Infant Development, 3rd ed. San Antonio, Tex: The Psychological Corporation; 2005

21. Moore V, Goodson S. How well does early diagnosis of autism stand the test of time?

22. Follow-up study of children assessed for autism at age 2 and development of an early diagnostic service. Autism. 2003;7:47-63. Abstract

23. Cox A, Klein K, Charman T, et al. Autism spectrum disorders at 20 and 42 months of age: stability of clinical and ADI-R diagnosis. J Child Psychol Psychiatry. 1999;40:719-732. Abstract

24. Stone WL, Lee EB, Ashford L, et al. Can autism be diagnosed accurately in children under 3 years? J Child Psychol Psychiatry. 1999;40:219-226.

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